

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: June 24, 2015

Subcommittee Members Attending

Barbara Burton, Lurie Children's Hospital – Chair
Darrel Waggoner, Lainie Friedman Ross, Maria Helgeson - University of Chicago
George Hoganson, Zohra Shad, Rich Dineen, Annie McRae- University of Illinois Chicago
Jennifer Burton - University of Illinois Chicago at Peoria
Michael Schneider- Carle Hospital-Champaign
David Dimmock-Medical College of Wisconsin
Tess Rhodes, DSCC

IDPH Staff

Rebecca Barnett, Khaja Basheeruddin, Jean Becker, Matt Charles, Maria Crain, David Culp, George Dizikes, Shannon Harrison, Tom Johnson, Heather Shryock

Background

The meeting was called to order at 4:05 p.m.

IDPH Laboratory Status Report

Khaja Basheeruddin reported that there have been over 10,000 specimens tested since the June 1 statewide implementation, and over 15,000 since the pilot began November 3, 2014. Since November, there have been 73 newborns with abnormal findings, totaling 89 specimens (49 presumptive positives, 36 borderline, 4 invalid). Below are the additional breakdowns of abnormal specimens:

LSD Type	Result	Pilot	Statewide	Total	Specimen Follow-up Status
Fabry					
	Invalid	1	0	1 (0 NICU)	1 normal
	Borderline	2	2	4 (3 NICU)	2 normal, 2 pending
	Positive	2	1	3 (0 NICU)	2 normal, 1 DX late onset
	Total	5 (0.03%)	3 (0.03%)	8 (0.03%)	
Gaucher					
	Invalid	0	1	1 (0 NICU)	1 pending
	Borderline	3	3	6 (5 NICU)	3 normal, 3 pending
	Positive	2	9	11 (9 NICU)	2 normal, 6 pending, 3 expired
	Total	5 (0.03%)	13 (0.13%)	18 (0.07%)	
MPS I					
	Invalid	0	0	0	
	Borderline	5	3	8 (2 NICU)	3 normal, 5 pending
	Positive	12	2	14 (11 NICU)	6 normal, 8 pending
	Total	17 (0.11%)	5 (0.05%)	22 (0.09%)	
Niemann-Pick					
	Invalid	0	0	0	
	Borderline	1	0	1 (1 NICU)	1 normal
	Positive	0	0	0	
	Total	1 (0.01%)	0	1 (<0.01%)	
Pompe					
	Invalid	2	0	2 (2 NICU)	2 pending
	Borderline	16	1	17 (12 NICU)	10 normal, 6 pending, 1 expired
	Positive	16	5	21 (15 NICU)	6 normal, 13 pending, 2 DX pseudo deficiency (same child)
	Total	34 (0.22%)	6 (0.06%)	40 (0.16%)	

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Compared to data in the pilot, the Pompe cutoff was fine tuned and cases have decreased, MPS 1 cases have decreased and Gaucher have increased. During the first week of state-wide testing the Perkin Elmer buffer containing an inhibitor for MPS 1 enzyme was used. This buffer was not helpful in Gaucher. As a result, the lab switched to the original buffer lacking the MPS1 inhibitor.

Dr. Basheeruddin reported invalid dried blood spots cannot be replicated. If repeat (3 punches) and initial analysis for specimens cannot be reproduced, CV >25%, the lab will repeat the analysis using seven punches. If the variation persists, the lab will call the specimen "Invalid". Furthermore, if multiple enzyme activities are below positive cut-off, the sample will be considered "Invalid".

In reviewing the data, the specialists agreed it may be important to include the following information: gestational age, NICU status, age at collection, TPN status. For gestational age, the recommendation was to break it out by: ≤ 32 weeks, 32-36 weeks, and ≥ 37 weeks. The question was raised if there was a positive newborn screen on an extremely pre-term baby, and the repeat was normal, was it appropriate to close the case if <32 weeks gestation. Dr. Burton said that more data was needed to make a protocol change and to check with the specialist for guidance for now.

The lab reported a fifth instrument will be installed in a couple of weeks. Additionally, the lab has received more material from Dr. Gelb to test MPS 2. Tom Johnson reported that the Krabbe contract had been sent to the University of Illinois today for review of changes to the contract. The specialists did individual case reviews of patients with consents on file. One NICU patient who tested positive for Gaucher and expired before additional testing could be done. The question was raised what advice they should tell the family about Gaucher. Both Drs. Hoganson and Burton agreed that since they were not able to confirm the diagnosis, they would not say anything as the likelihood was so remote.

Genetic Counselor Annie McRae discussed a case that due to the high cost of diagnostic work up, the family was uncomfortable moving forward. She said when the diagnostic enzyme tests are negative, and they are recommending molecular testing these tests can cost \$3,000 - \$5,000 and are often not covered. Rich Dineen reiterated that there are no back up programs for these families and co-pays are high.

Meeting adjourned at 4:50 p.m.